## SEQUENCE LISTING

<110> DENEFLE, Patrice ROSIER-MONTUS, Marie-Francoise PRADES, Catherine ARNOULD-REGUIGNE, Isabelle DUVERGER, Nicolas ALLIKMETS, Rando DEAN, Michael

<120> NUCLEIC ACIDS OF THE HUMAN ABCA5, ABCA6, ABCA9, AND ABCA10 GENES, VECTORS CONTAINING SUCH NUCLEIC ACIDS, AND USES THEREOF

<130> ABCA5, 6, 9, 10
<140> US 10/005,338
<141> 2001-12-07
<150> US 60/263,231
<151> 2001-01-23

<150> FR 00403440.1 <151> 2000-12-07

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<170> PatentIn Ver. 2.1

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Leu Arg Asn Leu Ser Phe Asp Ile Tyr Glu Gly Gln Ile Thr Ala Leu 500 505 510 Leu Gly His Ser Gly Thr Gly Lys Ser Thr Leu Met Asn Ile Leu Cys 515 520 525 Gly Leu Cys Pro Pro Ser Asp Gly Phe Ala Ser Ile Tyr Gly His Arg 530 540 Val Ser Glu Ile Asp Glu Met Phe Glu Ala Arg Lys Met Ile Gly Ile 545 550 555 560 Cys Pro Gln Leu Asp Ile His Phe Asp Val Leu Thr Val Glu Glu Asn 565 570 575 Leu Ser Ile Leu Ala Ser Ile Lys Gly Ile Pro Ala Asn Asn Ile Ile 580 585 590 Gln Glu Val Gln Lys Val Leu Leu Asp Leu Asp Met Gln Thr Ile Lys 595 600 605 Asp Asn Gln Ala Lys Lys Leu Ser Gly Gly Gln Lys Arg Lys Leu Ser 610 615 620 Leu Gly Ile Ala Val Leu Gly Asn Pro Lys Ile Leu Leu Leu Asp Glu 625 630 635 640 Pro Thr Ala Gly Met Asp Pro Cys Ser Arg His Ile Val Trp Asn Leu 645 650 655 Leu Lys Tyr Arg Lys Ala Asn Arg Val Thr Val Phe Ser Thr His Phe 660 670 Met Asp Glu Ala Asp Ile Leu Ala Asp Arg Lys Ala Val Ile Ser Gln 685 Gly Met Leu Lys Cys Val Gly Ser Ser Met Phe Leu Lys Ser Lys Trp  $690 \hspace{1.5cm} 695 \hspace{1.5cm} 700$ Gly Ile Gly Tyr Arg Leu Ser Met Tyr Ile Asp Lys Tyr Cys Ala Thr 705 710 715 720Glu Ser Leu Ser Ser Leu Val Lys Gln His Ile Pro Gly Ala Thr Leu 725 730 735 Leu Gln Gln Asn Asp Gln Gln Leu Val Tyr Ser Leu Pro Phe Lys Asp 740 745 750 Met Asp Lys Phe Ser Gly Leu Phe Ser Ala Leu Asp Ser His Ser Asn 765 760 765 Leu Gly Val Ile Ser Tyr Gly Val Ser Met Thr Thr Leu Glu Asp Val 770 775 780 Phe Leu Lys Leu Glu Val Glu Ala Glu Ile Asp Gln Ala Asp Tyr Ser 785 790 795 800 Val Phe Thr Gln Gln Pro Leu Glu Glu Met Asp Ser Lys Ser Phe 805 810 815 Asp Glu Met Glu Gln Ser Leu Leu Ile Leu Ser Glu Thr Lys Ala Ser 820 825 830

Leu Val Ser Thr Met Ser Leu Trp Lys Gln Gln Met Tyr Thr Ile Ala 840 Lys Phe His Phe Phe Thr Leu Lys Arg Glu Ser Lys Ser Val Arg Ser Val Leu Leu Leu Leu Ile Phe Phe Thr Val Gln Ile Phe Met Phe 865 870 875 880 865 Leu Val His His Ser Phe Lys Asn Ala Val Val Pro Ile Lys Leu Val Pro Asp Leu Tyr Phe Leu Lys Pro Gly Asp Lys Pro His Lys Tyr Lys 900 905 910 Thr Ser Leu Leu Gln Asn Ser Ala Asp Ser Asp Ile Ser Asp Leu
915 920 925 Ile Ser Phe Phe Thr Ser Gln Asn Ile Met Val Thr Met Ile Asn Asp 935 Ser Asp Tyr Val Ser Val Ala Pro His Ser Ala Ala Leu Asn Val Met 945 950 955 960 His Ser Glu Lys Asp Tyr Val Phe Ala Ala Val Phe Asn Ser Thr Met 965 970 975 val Tyr Ser Leu Pro Ile Leu Val Asn Ile Ile Ser Asn Tyr Tyr Leu 985 Tyr His Leu Asn Val Thr Glu Thr Ile Gln Ile Trp Ser Thr Pro Phe 1000 Phe Gln Glu Ile Thr Asp Ile Val Phe Lys Ile Glu Leu Tyr Phe Gln 1020 Ala Ala Leu Leu Gly Ile Ile Val Thr Ala Met Pro Pro Tyr Phe Ala 1030 1025 Met Glu Asn Ala Glu Asn His Lys Ile Lys Ala Tyr Thr Gln Leu Lys 1045 1050 1055 1045 Leu Ser Gly Leu Leu Pro Ser Ala Tyr Trp Ile Gly Gln Ala Val 1065 1060 Asp Ile Pro Leu Phe Phe Ile Ile Leu Ile Leu Met Leu Gly Ser Leu 1080 Leu Ala Phe His Tyr Gly Leu Tyr Phe Tyr Thr Val Lys Phe Leu Ala Val Val Phe Cys Leu Ile Gly Tyr Val Pro Ser Val Ile Leu Phe Thr 1115 1110 1105 Tyr Ile Ala Ser Phe Thr Phe Lys Lys Ile Leu Asn Thr Lys Glu Phe 1130 Trp Ser Phe Ile Tyr Ser Val Ala Ala Leu Xaa Cys Ile Ala Ile Thr 1145 1150 Glu Ile Thr Phe Phe Met Gly Tyr Thr Ile Ala Thr Ile Leu His Tyr 1160 1165 Ala Phe Cys Ile Ile Ile Pro Ile Tyr Pro Leu Leu Gly Cys Leu Ile Page 11

Ser Phe Ile Lys Ile Ser Trp Lys Asn Val Arg Lys Asn Val Asp Thr 1185 1190 1195 1200

Tyr Asn Pro Trp Asp Arg Leu Ser Val Ala Val Ile Ser Pro Tyr Leu 1205 1210 1215

Gln Cys Val Leu Trp Ile Phe Leu Leu Gln Tyr Tyr Glu Lys Lys Tyr 1220 1225 1230

Gly Gly Arg Ser Ile Arg Lys Asp Pro Phe Phe Arg Asn Leu Ser Thr 1235 1240 1245

Lys Ser Lys Asn Arg Lys Leu Pro Glu Pro Pro Asp Asn Glu Asp Glu 1250 1260

Asp Glu Asp Val Lys Ala Glu Arg Leu Lys Val Lys Glu Leu Met Gly 1265 1270 1280

Cys Gln Cys Cys Glu Glu Lys Pro Ser Ile Met Val Ser Asn Leu His 1285 1290 1295

Lys Glu Tyr Asp Asp Lys Lys Asp Phe Leu Leu Ser Arg Lys Val Lys 1300 1310

Lys Val Ala Thr Lys Tyr Ile Ser Phe Cys Val Lys Lys Gly Glu Ile 1315 1320 1325

Leu Gly Leu Leu Gly Pro Asn Gly Ala Gly Lys Ser Thr Ile Ile Asn 1330 1335 1340

Ile Leu Val Gly Asp Ile Glu Pro Thr Ser Gly Gln Val Phe Leu Gly 1345 1350 1355 1360

Asp Tyr Ser Ser Glu Thr Ser Glu Asp Asp Asp Ser Leu Lys Cys Met 1365 1370 1375

Gly Tyr Cys Pro Gln Ile Asn Pro Leu Trp Pro Asp Thr Thr Leu Gln 1380 1385 1390

Glu His Phe Glu Ile Tyr Gly Ala Val Lys Gly Met Ser Ala Ser Asp 1395 1400 1405

Met Lys Glu Val Ile Ser Arg Ile Thr His Ala Leu Asp Leu Lys Glu 1410 1415 1420

His Leu Gln Lys Thr Val Lys Lys Leu Pro Ala Gly Ile Lys Arg Lys 1425 1430 1440

Leu Cys Phe Ala Leu Ser Met Leu Gly Asn Pro Gln Ile Thr Leu Leu 1445 1450 1455

Asp Glu Pro Ser Thr Gly Met Asp Pro Lys Ala Lys Gln His Met Trp 1460 1465 1470

Arg Ala Ile Arg Thr Ala Phe Lys Asn Arg Lys Arg Ala Ala Ile Leu 1475 1480 1485

Thr Thr His Tyr Met Glu Glu Ala Glu Ala Val Cys Asp Arg Val Ala 1490 1495 1500

Ile Met Val Ser Gly Gln Leu Arg Cys Ile Gly Thr Val Gln His Leu 1505 1510 1520 Page 12 Lys Ser Lys Phe Gly Lys Gly Tyr Phe Leu Glu Ile Lys Leu Lys Asp 1525 1530 1535

Trp Ile Glu Asn Leu Glu Val Asp Arg Leu Gln Arg Glu Ile Gln Tyr 1540 1545 1550

Ile Phe Pro Asn Ala Ser Arg Gln Glu Ser Phe Ser Ser Ile Leu Ala 1555 1560 1565

Tyr Lys Ile Pro Lys Glu Asp Val Gln Ser Leu Ser Gln Ser Phe Phe 1570 1575 1580

Lys Leu Glu Glu Ala Lys His Ala Phe Ala Ile Glu Glu Tyr Ser Phe 1585 1590 1595 1600

Ser Gln Ala Thr Leu Glu Gln Val Phe Val Glu Leu Thr Lys Glu Gln 1605 1610 1615

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Glu Arg Thr Gln Glu Asp Arg Val Val Phe 1635 1640

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Ser Ser Met Arg Asn Val Gln Phe Pro Gly Met Ala Pro Gln Asn Leu 50 60

Gly Arg Val Asp Lys Phe Asn Ser Ser Ser Leu Met Val Val Tyr Thr 65 70 75 80

Pro Ile Ser Asn Leu Thr Gln Gln Ile Met Asn Lys Thr Ala Leu Ala 85 90 95

Pro Leu Lys Gly Thr Ser Val Ile Gly Ala Pro Asn Lys Thr His  $100 \hspace{1cm} 105 \hspace{1cm} 110$ 

Met Asp Glu Ile Leu Leu Glu Asn Leu Pro Tyr Ala Met Gly Ile Ile 115 120 125

Phe Asn Glu Thr Phe Ser Tyr Lys Leu Ile Phe Phe Gln Gly Tyr Asn 130 135 140

Gly Glu Phe Ser Cys Thr Leu Thr Lys Tyr Trp Asn Arg Gly Phe Val Page 13 165 170 175

Ala Leu Gln Thr Ala Ile Asn Thr Ala Ile Ile Glu Ile Thr Thr Asn Pro Val Met Glu Glu Leu Met Ser Val Thr Ala Ile Thr Met Lys 195 200 205 Thr Leu Pro Phe Ile Thr Lys Asn Leu Leu His Asn Glu Met Phe Ile 210 220 Leu Phe Phe Leu Leu His Phe Ser Pro Leu Val Tyr Phe Ile Ser Leu 225 230 235 240 Asn Val Thr Lys Glu Arg Lys Lys Ser Lys Asn Leu Met Lys Met Met 245 250 255 Gly Leu Gln Asp Ser Ala Phe Trp Leu Ser Trp Gly Leu Ile Tyr Ala 260 265 270 Gly Phe Ile Phe Ile Ile Ser Ile Phe Ile Thr Ile Ile Ile Thr Phe 275 280 285 Thr Gln Ile Ile Val Met Thr Gly Phe Met Val Ile Phe Ile Leu Phe 290 295 300 Phe Leu Tyr Gly Leu Ser Leu Val Ala Leu Val Phe Leu Met Ser Val Leu Leu Lys Lys Ala Val Leu Thr Asn Leu Val Val Phe Leu Leu Thr 325 330 335 Leu Phe Trp Gly Cys Leu Gly Phe Thr Val Phe Tyr Glu Gln Leu Pro 340 350 Ser Ser Leu Glu Trp Ile Leu Asn Ile Cys Ser Pro Phe Ala Phe Thr 355 360 365 Thr Gly Met Ile Gln Ile Ile Lys Leu Asp Tyr Asn Leu Asn Gly Val 370 375 380 Ile Phe Pro Asp Pro Ser Gly Asp Ser Tyr Thr Met Ile Ala Thr Phe 385 390 395 400 Ser Met Leu Leu Leu Asp Gly Leu Ile Tyr Leu Leu Leu Ala Leu Tyr 405 410 415 Phe Asp Lys Ile Leu Pro Tyr Gly Asp Glu Arg His Tyr Ser Pro Leu 420 425 430 Phe Phe Leu Asn Ser Ser Ser Cys Phe Gln His Gln Arg Thr Asn Ala 435 440 445 Lys Val Ile Glu Lys Glu Ile Asp Ala Glu His Pro Ser Asp Asp Tyr 450 460 Phe Glu Pro Val Ala Pro Glu Phe Gln Gly Lys Glu Ala Ile Arg Ile 465 470 475 480 Arg Asn Val Lys Lys Glu Tyr Lys Gly Lys Ser Gly Lys Val Glu Ala 485 490 495 Leu Lys Gly Leu Leu Phe Asp Ile Tyr Glu Gly Gln Ile Thr Ala Ile 500 505 510 Page 14

Leu Gly His Ser Gly Ala Gly Lys Ser Ser Leu Leu Asn Ile Leu Asn 515 520 525 Gly Leu Ser Val Pro Thr Glu Gly Ser Val Thr Ile Tyr Asn Lys Asn 530 540 Cys Pro Gln Phe Asn Val Gln Phe Asp Ile Leu Thr Val Lys Glu Asn 565 570 575 Leu Ser Leu Phe Ala Lys Ile Lys Gly Ile His Leu Lys Glu Val Glu 580 585 590 Gln Glu Val Gln Arg Ile Leu Leu Glu Leu Asp Met Gln Asn Ile Gln 595 600 605 Asn Leu Ala Lys His Leu Ser Glu Gly Gln Lys Arg Lys Leu Thr 610 620 Phe Gly Ile Thr Ile Leu Gly Asp Pro Gln Ile Leu Leu Leu Asp Glu Pro Thr Thr Gly Leu Asp Pro Phe Ser Arg Asp Gln Val Trp Ser Leu 645 650 655 Leu Arg Glu Arg Arg Ala Asp His Val Ile Leu Phe Ser Thr Gln Ser 660 670 Met Asp Glu Ala Asp Ile Leu Ala Asp Arg Lys Val Ile Met Ser Asn 675 680 685 Gly Arg Leu Lys Cys Ala Gly Ser Ser Met Phe Leu Lys Arg Arg Trp  $690 \hspace{1.5cm} 695 \hspace{1.5cm} 700$ Gly Leu Gly Tyr His Leu Ser Leu His Arg Asn Glu Ile Cys Asn Pro 715 720 Glu Gln Ile Thr Ser Phe Ile Thr His His Ile Pro Asp Ala Lys Leu 725 730 735 Lys Thr Glu Asn Lys Glu Lys Leu Val Tyr Thr Leu Pro Leu Glu Arg 740 745 750 Thr Asn Thr Phe Pro Asp Leu Phe Ser Asp Leu Asp Lys Cys Ser Asp 765 765 Gln Gly Val Thr Gly Tyr Asp Ile Ser Met Ser Thr Leu Asn Glu Val 770 775 780 Phe Met Lys Leu Glu Gly Gln Ser Thr Ile Glu Gln Asp Phe Glu Gln 785 790 795 800 Val Glu Met Ile Arg Asp Ser Glu Ser Leu Asn Glu Met Glu Leu Ala 805 810 815 His Ser Ser Phe Ser Glu Met Gln Thr Ala Val Ser Asp Met Gly Leu Trp Arg Met Gln Val Phe Ala Met Ala Arg Leu Arg Phe Leu Lys Leu 835 840

Lys	Arg 850	Gln	Thr	Lys	Val	Leu 855	Leu	Thr	Leu	Leu	Leu 860	val	Phe	Gly	Ile
Ala 865	Ile	Phe	Pro	Leu	11e 870	val	Glu	Asn	Ile	11e 875	Tyr	ΑΊа	Met	Leu	Asn 880
Glu	Lys	Ile	Asp	Trp 885	Glu	Phe	Lys	Asn	G]u 890	Leu	Tyr	Phe	Leu	Ser 895	Pro
Gly	Gln	Leu	Pro 900	Gln	Glu	Pro	Arg	Thr 905	Ser	Leu	Leu	Ile	Ile 910	Asn	Asn
Thr	Glu	Ser 915	Asn	Ile	Glu	Asp	Phe 920	Ile	Lys	Ser	Leu	Lys 925	His	Gln	Asn
Ile	Leu 930	Leu	Glu	val	Asp	Asp 935	Phe	Glu	Asn	Arg	Asn 940	Gly	Thr	Asp	Gly
Leu 945	Ser	Tyr	Asn	Gly	Ala 950	Ile	Ile	val	Ser	Gly 955	Lys	Gln	Lys	Asp	Tyr 960
Arg	Phe	Ser	۷a٦	va1 965	Cys	Asn	Thr	Lys	Arg 970	Leu	His	Cys	Phe	Pro 975	Ile
Leu	Met	Asn	Ile 980	Ile	Ser	Asn	Gly	Leu 985	Leu	Gln	Met	Phe	Asn 990	ніѕ	Thr
Gln	His	Ile 995	Arg	Ile	Glu		Ser LOOO	Pro	Phe	Pro		Ser LOO5	His	Ile	Gly
	Trp LO10	Thr	Gly	Leu		Asp LO15	Gly	Ser	Phe	Phe	Leu L020	Phe	Leu	Val	Leu
Cys 1025		Ile	Ser		Tyr L030	Ile	Thr	Met		Ser LO35	Ile	Ser	Asp	Tyr	Lys 1040
Lys	Asn	Ala		Ser LO45	Gln	Leu	Trp		Ser LO50	Gly	Leu	Tyr	Thr	Ser 1055	Ala
Tyr	Trp		G]y L060	Gln	Ala	Leu	val	Asp L065	٧a٦	Ser	Phe		11e 1070	Leu	Ile
Leu		Leu L075	Met	Tyr	Leu		Phe L080	Tyr	Ile	Glu		Met LO85	Gln	Tyr	Leu
	11e 1090	Thr	Ser	Gln			Phe	Ala	Leu	٧a٦	Ile	٧al	Thr	Pro	Gly
Tyr					-	1095					1100				
1109	Ala 5	Ala	Ser				Phe		Tyr		1100		Phe	Ile	Phe 1120
	5		Arg		∨a1 1110	Phe	Phe Gly	Ile Leu	Tyr :	Met 1115	Iloo Ile	Ser	Phe	-	1120
Arg	Lys	Arg Thr	Arg	Lys 1125	Val 1110 Asn	Phe	Gly	Ile Leu	Tyr E Trp 1130	Met 1115 Ser	IlOO Ile Phe	Ser Tyr His	Phe	Phe 1135	1120 Phe
Arg Ala	Lys Ser Ile	Arg Thr	Arg Ile 1140	Lys L125 Met	Val 1110 Asn Phe	Phe Ser Ser Met	Gly	Ile Leu Thr L145	Tyr Irp 1130 Leu	Met 1115 Ser Ile	Ile Phe Asn Ser	Ser Tyr His	Phe Phe 1150	Phe 1135 Asp	Phe Leu
Arg Ala Ser Gly	Lys Ser Ile	Arg Thr Leu 1155	Arg Ile 1140 Ile	Lys 1125 Met Thr	Val 1110 Asn Phe Thr	Phe Ser Ser Met	Gly Ile	Ile Leu Thr L145 Leu	Tyr Trp 1130 Leu Val	Met 1115 Ser Ile Pro	Iloo Ile Phe Asn Ser	Ser Tyr His Tyr 1165	Phe Phe 1150 Thr	Phe 1135 Asp Leu	Phe Leu Leu

Phe Ile Pro Tyr Phe Gln Thr Leu Leu Phe Val Phe Val Leu Arg Cys 1205 1210 1215

Met Glu Leu Lys Cys Gly Lys Lys Arg Met Arg Lys Asp Pro Val Phe 1220 1230

Arg Ile Ser Pro Gln Ser Arg Asp Ala Lys Pro Asn Pro Glu Glu Pro 1235 1240 1245

Ile Asp Glu Asp Glu Asp Ile Gln Thr Glu Arg Ile Arg Thr Ala Thr 1250 1255 1260

Ala Leu Thr Thr Ser Ile Leu Asp Glu Lys Pro Val Ile Ile Ala Ser 1265 1270 1275 1280

Cys Leu His Lys Glu Tyr Ala Gly Gln Lys Lys Ser Cys Phe Ser Lys 1285 1290 1295

Arg Lys Lys Ile Ala Ala Arg Asn Ile Ser Phe Cys Val Gln Glu 1300 1305 1310

Gly Glu Ile Leu Gly Leu Leu Gly Pro Ser Gly Ala Gly Lys Ser Ser 1315 1320 1325

Ser Ile Arg Met Ile Ser Gly Ile Thr Lys Pro Thr Ala Gly Glu Val 1330 1335 1340

Glu Leu Lys Gly Cys Ser Ser Val Leu Gly His Leu Gly Tyr Cys Pro 1345 1350 1355 1360

Gln Glu Asn Val Leu Trp Pro Met Leu Thr Leu Arg Glu His Leu Glu 1365 1370 1375

Val Tyr Ala Ala Val Lys Gly Leu Arg Lys Ala Asp Ala Arg Leu Ala 1380 1385 1390

Ile Ala Arg Leu Val Ser Ala Phe Lys Leu His Glu Gln Leu Asn Val 1395 1400 1405

Pro Val Gln Lys Leu Thr Ala Gly Ile Thr Arg Lys Leu Cys Phe Val 1410 1415 1420

Leu Ser Leu Leu Gly Asn Ser Pro Val Leu Leu Leu Asp Glu Pro Ser 1425 1430 1435 1440

Thr Gly Ile Asp Pro Thr Gly Gln Gln Met Trp Gln Ala Ile Gln
1445 1450 1455

Ala Val Val Lys Asn Thr Glu Arg Gly Val Leu Leu Thr Thr His Asn 1460 1465 1470

Leu Ala Glu Ala Glu Ala Leu Cys Asp Arg Val Ala Ile Met Val Ser 1475 1480 1485

Gly Arg Leu Arg Cys Ile Gly Ser Ile Gln His Leu Lys Asn Lys Leu 1490 1495 1500

Gly Lys Asp Tyr Ile Leu Glu Leu Lys Val Lys Glu Thr Ser Gln Val 1505 1510 1515 1520

Thr Leu Val His Thr Glu Ile Leu Lys Leu Phe Pro Gln Ala Ala Gly 1525 1530 1535 Page 17 Gln Glu Arg Tyr Ser Ser Leu Leu Thr Tyr Lys Leu Pro Val Ala Asp 1540

Val Tyr Pro Leu Ser Gln Thr Phe His Lys Leu Glu Ala Val Lys His 1565

Asn Phe Asn Leu Glu Glu Tyr Ser Leu Ser Gln Cys Thr Leu Glu Lys 1570

Val Phe Leu Glu Leu Ser Lys Glu Gln Glu Val Gly Asn Phe Asp Glu 1585

Glu Ile Asp Thr Thr Met Arg Trp Lys Leu Leu Pro His Ser Asp Glu 1605

Pro

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Met Lys Ile Leu Pro Phe Val Ala Gln Gly Gly Val Ala Thr Asp 210 215 220 Phe Phe Ile Phe Phe Cys Ile Ile Ser Phe Ser Thr Phe Ile Tyr Tyr 225 230 235 240 Val Ser Val Asn Val Thr Gln Glu Arg Gln Tyr Ile Thr Ser Leu Met 245 250 255 Thr Met Met Gly Leu Arg Glu Ser Ala Phe Trp Leu Ser Trp Gly Leu 260 265 270 Met Tyr Ala Gly Phe Ile Leu Ile Met Ala Thr Leu Met Ala Leu Ile 275 280 285 Val Lys Ser Ala Gln Ile Val Val Leu Thr Gly Phe Val Met Val Phe 290 295 300 Thr Leu Phe Leu Leu Tyr Gly Leu Ser Leu Ile Thr Leu Ala Phe Leu 305 310 315 320 Met Ser Val Leu Ile Lys Lys Pro Phe Leu Thr Gly Leu Val Val Phe 325 330 335 Leu Leu Ile Val Phe Trp Gly Ile Leu Gly Phe Pro Ala Leu Tyr Thr 340 345 350 His Leu Pro Ala Phe Leu Glu Trp Thr Leu Cys Leu Leu Ser Pro Phe 355 360 365 Ala Phe Thr Val Gly Met Ala Gln Leu Ile His Leu Asp Tyr Asp Val 370 375 380 Asn Ser Asn Ala His Leu Asp Ser Ser Gln Asn Pro Tyr Leu Ile Ile 385 390 395 400 Ala Thr Leu Phe Met Leu Val Phe Asp Thr Leu Leu Tyr Leu Val Leu 405 410 415Thr Leu Tyr Phe Asp Lys Ile Leu Pro Ala Glu Tyr Gly His Arg Cys 420 425 430 Ser Pro Leu Phe Phe Leu Lys Ser Cys Phe Trp Phe Gln His Gly Arg 435 440 445 Ala Asn His Val Val Leu Glu Asn Glu Thr Asp Ser Asp Pro Thr Pro 450 455 460 Asn Asp Cys Phe Glu Pro Val Ser Pro Glu Phe Cys Gly Lys Glu Ala 465 470 475 480 Ile Arg Ile Lys Asn Leu Lys Lys Glu Tyr Ala Gly Lys Cys Glu Arg 485 490 495 Val Glu Ala Leu Lys Gly Val Val Phe Asp Ile Tyr Glu Gly Gln Ile 500 505 510 Thr Ala Leu Leu Gly His Ser Gly Ala Gly Lys Thr Thr Leu Leu Asn 515 520 525 Ile Leu Ser Gly Leu Ser Val Pro Thr Ser Gly Ser Val Thr Val Tyr 535

Asn His Thr Leu Ser Arg Met Ala Asp Ile Glu Asn Ile Ser Lys Phe 545 550 555 560 Thr Gly Phe Cys Pro Gln Ser Asn Val Gln Phe Gly Phe Leu Thr Val 565 570 575 Lys Glu Asn Leu Arg Leu Phe Ala Lys Ile Lys Gly Ile Leu Pro His 580 585 590 Glu Val Glu Lys Glu Val Gln Arg Val Val Gln Glu Leu Glu Met Glu 595 600 605 Asn Ile Gln Asp Ile Leu Ala Gln Asn Leu Ser Gly Gly Gln Asn Arg 610 620 Lys Leu Thr Phe Gly Ile Ala Ile Leu Gly Asp Pro Gln Val Leu Leu 625 630 635 640 Leu Asp Glu Pro Thr Ala Gly Leu Asp Pro Leu Ser Arg His Arg Ile 645 650 655 Trp Asn Leu Leu Lys Glu Gly Lys Ser Asp Arg Val Ile Leu Phe Ser 660 665 670 Thr Gln Phe Ile Asp Glu Ala Asp Ile Leu Ala Asp Arg Lys Val Phe 675 680 685 Ile Ser Asn Gly Lys Leu Lys Cys Ala Gly Ser Ser Leu Phe Leu Lys 690 695 700 Lys Lys Trp Gly Ile Gly Tyr His Leu Ser Leu His Leu Asn Glu Arg 705 710 715 720 Cys Asp Pro Glu Ser Ile Thr Ser Leu Val Lys Gln His Ile Ser Asp 725 730 735 Ala Lys Leu Thr Ala Gln Ser Glu Glu Lys Leu Val Tyr Ile Leu Pro 740 745 750 Leu Glu Arg Thr Asn Lys Phe Pro Glu Leu Tyr Arg Asp Leu Asp Arg 765 760 765 Ser Asn Gln Gly Ile Glu Asp Tyr Gly Val Ser Ile Thr Thr Leu 770 780 Asn Glu Val Phe Leu Lys Leu Glu Gly Lys Ser Thr Ile Asp Glu Ser 785 790 795 800 Asp Ile Gly Ile Trp Gly Gln Leu Gln Thr Asp Gly Ala Lys Asp Ile 805 810 815 Gly Ser Leu Val Glu Leu Glu Gln Val Leu Ser Ser Phe His Glu Thr 820 825 830 Arg Lys Thr Ile Ser Gly Val Ala Leu Trp Arg Gln Gln Val Cys Ala 835 840 845 Ile Ala Lys Val Arg Phe Leu Lys Leu Lys Lys Glu Arg Lys Ser Leu 850 860 Trp Thr Ile Leu Leu Phe Gly Ile Ser Phe Ile Pro Gln Leu Leu 865 870 875 880 870

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Ser	Pro	Asn	Thr 900	Tyr	Phe	Leu	Ser	Pro 905	Gly	Gln	Gln	Pro	Gln 910	Asp	Pro
Leu	Thr	ніs 915	Leu	Leu	۷al	Ile	Asn 920	Lys	Thr	Gly	Ser	Thr 925	Ile	Asp	Asn
Phe	Leu 930	His	Ser	Leu	Arg	Arg 935	Gln	Asn	Ile	Аla	11e 940	Glu	٧a٦	Asp	Ala
Phe 945	Gly	Thr	Arg	Asn	Gly 950	Thr	Asp	Asp	Pro	Ser 955	Tyr	Asn	Gly	Ala	11e 960
Ile	val	Ser	Gly	Asp 965	Glu	Lys	Asp	ніѕ	Arg 970	Phe	Ser	Ile	Ala	Cys 975	Asn
Thr	Lys	Arg	Leu 980	Asn	Cys	Phe	Pro	Val 985	Leu	Leu	Asp	val	Ile 990	Ser	Asn
Glу	Leu	Leu 995	Gly	Ile	Phe	Asn 1	Ser LOOO	Ser	Glu	His		G1n L005	Thr	Asp	Arg
	Thr L010	Phe	Phe	Glu		His 1015	Met	Asp	Tyr		Tyr L020	Gly	Tyr	Arg	Ser
Asn 1025		Phe	Phe		1]e 1030	Pro	Met	Ala		Ser 1035	Phe	Thr	Pro	Tyr ]	11e 1040
Ala	Met	Ser		Ile LO45	Gly	Asp	Tyr	Lys	Lys 1050	Lys	Ala	His	Ser	G]n L055	Leu
Arg	Ile		G]y L060	Leu	Tyr	Pro		Ala 1065	Tyr	Trp	Phe		G1n L070	Ala	Leu
٧a٦		Va] 1075	Ser	Leu	Tyr		Leu L080	Ile	Leu	Leu	Leu 1	Met LO85	Gln	Ile	Met
	Tyr 1090	Ile	Phe	Ser		G]u L095	Glu	Ile	Ile	Phe	Ile L100	Ile	Gln	Asn	Leu
Leu 110		Gln	Ile		Cys L110	ser	Ile	Gly	Tyr :	Val 1115	ser	Ser	Leu	val :	Phe L120
Leu	Thr	Tyr	۷al	11e 1125	Ser	Phe	Ile	Phe	Arg 1130	Asn	Gly	Arg	Lys 1	Asn L135	Ser
Gly	Ile		Ser L140	Phe	Phe	Phe		11e 1145	val	val	Ile	Phe	Ser 1150	Ile	val
Ala	Thr	Asp 1155	Leu	Asn	Glu	Tyr :	Gly 1160	Phe	Leu	Gly	Leu	Phe L165	Phe	Gly	Thr
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Glu 118		Ser	Pro		Ser 1190	Met	Asp	Tyr	Leu	Gly 1195	Ala	Ser	Glu	Ser :	G]u 1200
Ile	۷a٦	туг		Ala 1205	Leu	Leu	Ile	Pro	Tyr 1210	Leu	His	Phe	Leu	Ile L215	Phe
Leu	Phe	Ile	Leu	Arg	Cys	Leu	Glu	Met	Asn P	Cys age	Arg 21	Lys	Lys	Leu	Met

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1660	4663	1230

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1445 1450 1455 Gln Gln Met Trp Gln Val Ile Arg Ala Thr Phe Arg Asn Thr Glu Arg Gly Ala Leu Leu Thr Thr His Tyr Met Ala Glu Ala Glu Ala Val Cys 1475 1480 1485 Asp Arg Val Ala Ile Met Val Ser Gly Arg Leu Arg Cys Ile Gly Ser 1490 1495 1500 Ile Gln His Leu Lys Ser Lys Phe Gly Lys Asp Tyr Leu Leu Glu Met 1505 1510 1515 1520 Lys Leu Lys Asn Leu Ala Gln Met Glu Pro Leu His Ala Glu Ile Leu 1530 Arg Leu Phe Pro Gln Ala Ala Gln Gln Glu Arg Phe Ser Ser Leu Met Val Tyr Lys Leu Pro Val Glu Asp Val Arg Pro Leu Ser Gln Ala Phe 1555 1560 1565 Page 22

Phe Lys Leu Glu Ile Val Lys Gln Ser Phe Asp Leu Glu Glu Tyr Ser 1570 1575 1580

Leu Ser Gln Ser Thr Leu Glu Gln Val Phe Leu Glu Leu Ser Lys Glu 1585 1590 1595 1600

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Gly Phe Met Val Ile Phe Thr Leu Tyr Ser Leu Tyr Gly Leu Ser Leu 210 220 Ile Ala Leu Ala Phe Leu Met Ser Val Leu Ile Arg Lys Pro Met Leu 225 230 235 240 Ala Gly Leu Ala Gly Phe Leu Phe Thr Val Phe Trp Gly Cys Leu Gly 245 250 255 Phe Thr Val Leu Tyr Arg Gln Leu Pro Leu Ser Leu Gly Trp Val Leu 260 265 270 Ser Leu Leu Ser Pro Phe Ala Phe Thr Ala Gly Met Ala Gln Val Thr 275 280 285 His Leu Asp Asn Tyr Leu Ser Gly Val Ile Phe Pro Asp Pro Ser Gly 290 295 300 Asp Ser Tyr Lys Met Ile Ala Thr Phe Phe Ile Leu Ala Phe Asp Thr 305 310 315 320Leu Phe Tyr Leu Ile Phe Thr Leu Tyr Phe Glu Arg Val Leu Pro Asp 325 330 335 Lys Asp Gly His Gly Asp Ser Pro Leu Phe Phe Leu Lys Ser Ser Phe 340 345 350 Trp Ser Lys His Gln Asn Thr His His Glu Ile Phe Glu Asn Glu Ile 355 360 365 Asn Pro Glu His Ser Ser Asp Asp Ser Phe Glu Pro Val Ser Pro Glu 370 375 380 Phe His Gly Lys Glu Ala Ile Arg Ile Arg Asn Val Ile Lys Glu Tyr 385 390 395 400 Asn Gly Lys Thr Gly Lys Val Glu Ala Leu Gln Gly Ile Phe Phe Asp 405 410 415 Ile Tyr Glu Gly Gln Ile Thr Ala Ile Leu Gly His Asn Gly Ala Gly 420 425 430 Lys Ser Thr Leu Leu Asn Ile Leu Ser Gly Leu Ser Val Ser Thr Glu 435 440 445 Gly Ser Ala Thr Ile Tyr Asn Thr Gln Leu Ser Glu Ile Thr Asp Met 450 455 460 Glu Glu Ile Arg Lys Asn Ile Gly Phe Cys Pro Gln Phe Asn Phe Gln 465 470 475 480 Phe Asp Phe Leu Thr Val Arg Glu Asn Leu Arg Val Phe Ala Lys Ile 485 490 495 Lys Gly Ile Gln Pro Lys Glu Val Glu Gln Glu Val Lys Arg Ile Ile 500 505 510 Met Glu Leu Asp Met Gln Ser Ile Gln Asp Ile Ile Ala Lys Lys Leu 515 520 525 Ser Gly Gly Gln Lys Arg Lys Leu Thr Leu Gly Ile Ala Ile Leu Gly 530 540 Asp Pro Gln Val Leu Leu Asp Glu Pro Thr Ala Gly Leu Asp Pro 545 Phe Ser Arg His Arg Val Trp Ser Leu Leu Lys Glu His Lys Val Asp 565 570 575 Arg Leu Ile Leu Phe Ser Thr Gln Phe Met Asp Glu Ala Asp Ile Leu 580 585 590 Ala Asp Arg Lys Val Phe Leu Ser Asn Gly Lys Leu Lys Cys Ala Gly 595 600 605 Ser Ser Leu Phe Leu Lys Arg Lys Trp Gly Ile Gly Tyr His Leu Ser 610 620 Leu His Arg Asn Glu Met Cys Asp Thr Glu Lys Ile Thr Ser Leu Ile 625 630 635 640 Lys Gln His Ile Pro Asp Ala Lys Leu Thr Thr Glu Ser Glu Glu Lys 645 650 655 Leu Val Tyr Ser Leu Pro Leu Glu Lys Thr Asn Lys Phe Pro Asp Leu 660 670 Tyr Ser Asp Leu Asp Lys Cys Ser Asp Gln Gly Ile Arg Asn Tyr Ala 675 680 685 Ser Val Thr Ser Leu Asn Glu Val Phe Leu Asn Leu Glu Gly Lys 690 695 700 Ser Ala Ile Asp Glu Pro Asp Phe Asp Ile Gly Lys Gln Glu Lys Ile 705 710 715 720 His Val Thr Arg Asn Thr Gly Asp Glu Ser Glu Met Glu Gln Val Leu 725 730 735 Cys Ser Leu Pro Glu Thr Arg Lys Ala Val Ser Ser Ala Ala Leu Trp 740 745 750 Arg Gln Ile Tyr Ala Val Ala Thr Leu Arg Phe Leu Lys Leu Arg 755 760 765 Glu Arg Arg Ala Leu Leu Cys Leu Leu Leu Val Leu Gly Ile Ala 770 780 Phe Ile Pro Ile Ile Leu Glu Lys Ile Met Tyr Lys Val Thr Arg Glu 785 790 795 800 Thr His Cys Trp Glu Phe Ser Pro Ser Met Tyr Phe Leu Ser Leu Glu 805 810 815 Gln Ile Pro Lys Thr Pro Leu Thr Ser Leu Leu Ile Val Asn Asn Thr 820 825 830 Gly Ser Asn Ile Glu Asp Leu Val His Ser Leu Lys Cys Gln Asp Ile 835 840 845 Leu Glu Ile Asp Asp Phe Arg Asn Arg Asn Gly Ser Asp Asp Pro 850 860 Ser Tyr Asn Gly Ala Ile Ile Val Ser Gly Asp Gln Lys Asp Tyr Arg 865 870 875 880 Phe Ser Val Ala Cys Asn Thr Lys Lys Leu Asn Cys Phe Pro Val Leu Page 25

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Leu	His	Lys		Tyr 1205	Tyr	Glu	Thr	Lys	Lys 1210	Ser	Cys	Phe	Ser	Thr 1215	Arg
Lys	Lys		11e 1220	Ala	Ile	Arg		∨al 1225		Phe age		val	Lys 1230	Lys	Gly

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Ile Lys Met Ile Thr Gly Cys Thr Lys Pro Thr Ala Gly Val Val 1250 1260

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Lys Phe Leu Gly Tyr Cys Pro Gln Glu Asn Ser Leu Trp Pro Lys Leu 1285 1290 1295

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Lys Leu Glu Ala Met Lys Gln Thr Phe Asn Leu Glu Glu Tyr Ser Leu 1490 1495 1500

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gatactaatt atgttattgc attigcacct gaaiccaaaa ctacccaaga gaiaatgaac
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<212> DNA
<213> Homo sapiens
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<212> DNA
<213> Homo sapiens
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<213> Homo sapiens
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tctcctctat ggcctgtctt tg
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<212> DNA
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cctgcatttt tggaatggac tttgtgtctt cttagcccct ttgccttcac tgttgggatg 180
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gcccag
<210> 56
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<211> 111
<212> DNA
<213> Homo sapiens
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aatctēctga aāgaggggaa ātcagacāga gtāāttetet teageacēca gtītatagāī 120
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gaggctgaca ttctggcgg
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agaagaaatg gggcataggc taccatttaa g
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<212> DNA
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aaggacaaac aaatttccag
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<211> 120
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<212> DNA
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tctggaggca gcaggtctgt gcaatagcaa aagttcgctt cctaaagtta aagaaagaaa 180
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acaaccacag gatcctctga cccatttact ggtcatcaat aagacag
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<211> 134
<212> DNA
<213> Homo sapiens
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atgcctttgg aactagaaat ggcacagatg acccatctta caatggtgct atcattgtgt 120
                                       Page 37
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<212> DNA
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gagaatgcgc tgtggcccaa cctgacagtg aggcagcacc tggaggtgta cgctgccgtg 120
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<210> 80
<211> 76
<212> DNA
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gggaataaag cgaaag
<210> 81
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accgggatgg accccgaggg gcagcagcaa atgtg
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<212> DNA
<213> Homo sapiens
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catggcagag gctgaggcgg tgtgtgaccg agtggccatc atggtgtcag gaaggctgag 120
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<212> DNA
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ccaggctgct cagcaggaaa g
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<212> DNA
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tttcttcaaa ttagagatag
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<212> DNA
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accctattga cttccgggta aggggagtca attgattacc cagcagcaca gtatttgctt 540
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atagcaaaga cttggaacca accaaaatac ccacaaatga tagaccggat aaagaaaacg 780 tgacacatat acaccatgga atactatgca gccatagaaa aggatgagtt catattcttc 840
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taacatgtaa ataggcatta atttttgaga aatagaaatg tttatcctta atgtatttt 180
aatttgctaa cattgatttt ttattttctt tcctgaaata gcttatttcc taaaatgaaa 240
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<213> Homo sapiens
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cacagaccat ggcttttcag aagccaagct gaataaaaac agttttaaaa gaggcaacca 180
tttgtagagg agtccttgaa ggattctica itgttttctt ggacaaaaag agaccagtgg 240
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<210> 89
<211> 141
<212> DNA
<213> Homo sapiens
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agataagcgt gcgtcaacaa attcaggctc ttctgtacaa gaattttctt aaaaaatgga 120
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gaataaaaag agagtttatt g
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<211> 205
<212> DNA
<213> Homo sapiens
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gctacccgtt ttcctgaaca acctcctaaa gtcctgggaa gcgtggatca gtttaatgac 120 tctggcctgg tagtggcata tacaccagtc agtaacataa cacaaaggat aatgaataag 180
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<213> Homo sapiens
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ggggatatag aatcccagtt ataaaggagc actctgaata cacag
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<212> DNA
<213> Homo sapiens
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<212> DNA
<213> Homo sapiens
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ataccacctt tcatttctaa gggagaaatt atgaatgaat ggtttcattt tacttgctta 120
gtttctttct cttctttat atactttgca tcattaaatg ttgcaaggga aagaggaaaa 180
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<210> 94
<211> 142
<212> DNA
<213> Homo sapiens
<220>
<221> unsure
<222> 11
<223> n=unknown, may be a or g or c or t
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tagcttatat ggcctttctt tg
<210> 95
<211> 186
<212> DNA
<213> Homo sapiens
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cctttatctt tgggatgggt attaagtctt cttagccctt ttgccttcac tgctggaatg 180
                                                                    186
gcccag
<210> 96
<211> 148
<212> DNA
<213> Homo sapiens
<400> 96
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tacaaaatga tagccacttt tttcattttg gcatttgata ctcttttcta tttgatattc 120
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                                                                   169
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<212> DNA
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                                                                   111
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<211> 176
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<211> 139
<212> DNA
<213> Homo sapiens
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gaggctgaca tcttggctg
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<210> 105
<211> 120
<212> DNA
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<211> 199
<212> DNA
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ggagagctct tttgtgttt
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<210> 108
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<212> DNA
<213> Homo sapiens
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ātgactttag aāacāgaaat ģgctcagatg atccctccta caatggagcc atcatagtgt 120
                                        Page 44
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<212> DNA
<213> Homo sapiens
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<211> 118
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<213> Homo sapiens
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<212> DNA
<213> Homo sapiens
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<210> 119
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<212> DNA
<213> Homo sapiens
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<211> 76
<212> DNA
<213> Homo sapiens
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gggaataaag agaaag
<210> 121
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<213> Homo sapiens
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accgggatgg accccgaggg gcagcagcaa atgtg
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<212> DNA
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<212> DNA
<213> Homo sapiens
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acaggctgct tggcaggaaa g
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<212> DNA
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